Clinical Information from Published Studies

	Dominant, Sporadic N=31	Dominant, Familial N=10	Recessive, N=25	Total N=66
Unique variants	19	5	7	31
Mean age years (SD)	11(8.5)	40 (22.3)	27(16.1)	
Sex	M: 12, F: 19	M: 6, F: 4	M: 12, F: 13	M: 30, F: 36
Developmental delay	100% (31/31)	0% (0/10)	16% (4/25)	53% (35/66)
Hypotonia	39% (12/31)	0% (0/10)	16% (4/25)	30% (20/66)
Spasticity	77% (24/31)	100% (10/10)	100% (25/25)	89% (59/66)
Seizures	29% (9/31)	0% (0/10)	0% (0/25)	14% (9/66)
Any optic findings Optic nerve change	74% (23/31) 58% (18/31)	10% (1/10) 0% (0/10)	12% (3/25) 0% (0/25)	41% (27/66) 27%(18/66)
Abnormal MRI Cerebellar atrophy	86% (25/29) 75% (22/29)	25% (1/4) 0% (0/4)	36% (4/11) 18% (2/11)	68% (30/44) 55% (24/44)
Sensory neuropathy	48% (15/31)	0% (0/10)	36% (9/25)	55% (24/66)